



OPA3 gene

OPA3, outer mitochondrial membrane lipid metabolism regulator

Normal Function

The *OPA3* gene provides instructions for making a protein whose exact function is unknown. The OPA3 protein is found in structures called mitochondria, which are the energy-producing centers of cells. Researchers speculate that the OPA3 protein is involved in regulating the shape of mitochondria.

Health Conditions Related to Genetic Changes

Costeff syndrome

At least three mutations in the *OPA3* gene have been found to cause Costeff syndrome. This condition is characterized by vision loss, movement problems, and intellectual disability. Costeff syndrome is caused by mutations in both copies of the *OPA3* gene in each cell, leading to a loss of OPA3 protein function. An *OPA3* gene mutation that causes Costeff syndrome in the Iraqi Jewish population (written as 143-1G>C) alters the way the *OPA3* gene's instructions are put together to make the protein, which results in a lack of functional protein. Cells without any functional OPA3 protein have abnormally shaped mitochondria. These cells likely have reduced energy production and die sooner than normal, decreasing energy availability in the body's tissues. It is unclear why cells that control vision and movement are particularly affected.

other disorders

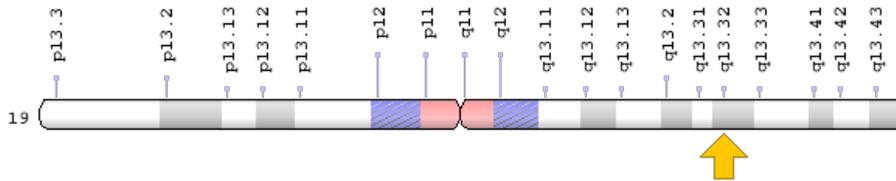
OPA3 gene mutations can also cause autosomal dominant optic atrophy and cataract (ADOAC). People with this condition have degeneration (atrophy) of the optic nerves, which carry information from the eyes to the brain, and clouding of the lens of the eye (cataract). These vision problems often occur in both eyes and can begin anytime from childhood to early adulthood. Some affected individuals have additional features such as hearing loss or movement problems.

People with ADOAC have a mutation in one copy of the *OPA3* gene in each cell. The mutations that cause ADOAC likely result in the production of an abnormal OPA3 protein that interferes with the functions of the cell. These cells likely have decreased energy production, leading to early cell death. It is unclear why cells within the eyes are particularly affected.

Chromosomal Location

Cytogenetic Location: 19q13.32, which is the long (q) arm of chromosome 19 at position 13.32

Molecular Location: base pairs 45,527,767 to 45,584,864 on chromosome 19 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- FLJ22187
- FLJ25932
- MGA3
- MGC75494
- OPA3 protein
- OPA3_HUMAN
- optic atrophy 3 (autosomal recessive, with chorea and spastic paraplegia)

Additional Information & Resources

Educational Resources

- Molecular Biology of the Cell (fourth edition, 2002): The Mitochondrion <https://www.ncbi.nlm.nih.gov/books/NBK26894/>
- The Cell: A Molecular Approach (second edition, 2000): Mitochondria <https://www.ncbi.nlm.nih.gov/books/NBK9896/>

GeneReviews

- OPA3-Related 3-Methylglutaconic Aciduria <https://www.ncbi.nlm.nih.gov/books/NBK1473>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28OPA3%5BTIAB%5D%29+OR+%28optic+atrophy+3%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- OPA3 GENE
<http://omim.org/entry/606580>
- OPTIC ATROPHY 3, AUTOSOMAL DOMINANT
<http://omim.org/entry/165300>

Research Resources

- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=OPA3%5Bgene%5D>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=8142
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/80207>
- UniProt
<http://www.uniprot.org/uniprot/Q9H6K4>

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